Supplemental materials:

Introduction:

Case reports of the 3q29 deletion, many of which are reviewed in[1], include[2-23].

Supplemental methods:

Neuroimaging Methods: MRI data were collected from a subset of study subjects (n=24) on a Siemens Magnetom Prisma 3T scanner at the Center for Systems Imaging Core using a 32-channel head coil. T1-weighted and T2-weighted high-resolution structural images were acquired. T1-weighted images were acquired using a 3D MPRAGE sequence with the following parameters: TE=2.24ms, TI=1000ms, TR=2400ms, flip angle=8^{o}, matrix=320x320, FOV=256x256mm, 208 sagittal slices, 0.8mm isotropic resolution, bandwidth=210 Hz/pixel. A GRAPPA factor of 2 was used with no phase oversampling and two repetitions. The total scan duration was 13 minutes, 16 seconds. A 3D T2-weighted Sampling Perfection with Application optimized Contrast using different angle Evolutions (SPACE) sequence was collected with the following parameters: TE=563ms, TR=3200ms, bandwidth=745 Hz/pixel, FOV=256×240×256 mm^3, sagittally acquired, 0.8mm isotropic resolution. A GRAPPA factor of 2 was used with no phase oversampling and two repetitions. The total scan duration was 11 minutes. Of the eight subjects who did not complete the MRI, two were at too low a developmental level to successfully complete the procedure, one declined to participate, and five were medically ineligible.

Supplemental results:

General Psychopathology:

In addition to the results reported in the text and Figure 2: two people met criteria for intermittent explosive disorder; two people met criteria for obsessive compulsive disorder; two people met criteria for major depressive disorder. For each of the following diagnoses, one person met criteria: oppositional defiant disorder, conduct disorder, Bipolar I disorder.

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Supplemental Tables:

Supplemental Table S1. Summary of study measures by domain

Evaluation	Domain & Measures
Medical	
	Medical History • Patient medical history by organ system • Family medical history • Family pedigree
	Physical Examination • Anthropomorphic Measures • Physical examination by organ system • Growth parameters (e.g., height, weight, head circumference)
Neurodevelopmental	
	Autism Spectrum Disorder • *Autism Diagnostic Interview -Revised* (ADI-R) • *Autism Diagnostic Observation Schedule, 2nd ed* (ADOS-2)
	Cognitive Ability & Adaptive Function • Differential Ability Scales, 2nd ed (DAS-II)[a] • Beery-Buktenica Developmental Test of Visual Motor Integration Test-6th ed (VMI-6) • *Behavioral Rating Inventory of Executive Functions, 2nd ed* (BRIEF-2)[a] and Adult version (BRIEF-A)[b] • Vineland Adaptive Behavior Scales, 3rd ed, Parent/Caregiver Form • Wechsler Abbreviated Scale of Intelligence, 2nd ed (WASI-II)[b]
Psychiatric	
	Anxiety • *Anxiety Disorders Interview Schedule for DSM –IV (ADIS-IV) Child Version*[a] • *Structured Clinical Interview for DSM-V --Research Version* (SCID-5-RV) - Module F[b, c]
	Prodromal Symptoms & Psychosis

	• Scheduled Interview for Psychosis-Risk Syndromes (SIPS) • *Structured Clinical Interview for DSM-V --Research Version* (SCID-5-RV) - Module B/C[b]
	General Psychopathology • *Kiddie Schedule for Affective Disorders and Schizophrenia* (K-SADS)[a, c] • *Structured Clinical Interview for DSM-V --Research Version* (SCID-5-RV) - Modules A, D, G, H, I, K[b]
Neuroimaging	Structural MRI
Neurological	Gross and fine motor skill assessment

[a] Administered to ages 6-18 years (, [b] administered to ages 19+ years, [c] For some cases ages 19-22 years, the K-SADS was used to assess anxiety and psychopathology.

Supplemental Table S2. Frequency of Symptoms and Diagnoses with Associated Procedure or Intervention (N = 32)

Symptoms/Diagnoses with Associated Procedure or Intervention	Associated HPO Codes	n	%
General		**7**	**22%**
Fatigue	HP:0012378	7	22%
HEENT		25	78%
Recurrent Infection addressed with surgery (any)		6	-
Cases requiring tonsillectomy	-	*4*	-
Cases requiring adenoidectomy	-	*5*	-
Eye (any)		**19**	**59%**
Brown Disease		1	3%
Prematurity Retinopathy	HP:0500049	1	3%
Glasses – unspecified reason	-	3	9%
Hypermetropia	HP:0000540	3	9%
Astigmatism	HP:0000483	5	16%
Myopia	HP:0000545	5	16%
Strabismus	HP:0000486	9	28%
Cases with strabismus requiring surgery		*3*	-
Ear			
Recurrent ear infection		**7**	**22%**
Cases requiring myringotomy and tube placement	**-**	*3*	**-**
Nose (any)		**8**	**25%**
Deviated Septum (*corrected by surgery*)	HP:0004411	1	3%
Epistaxis	HP:0000421	7	22%
Cases with epistaxis requiring surgery		*2*	-
Pharynx (any)		**4**	**13%**
Esophageal Dysmotility	HP:0031857	1	3%
Laryngomalacia	HP:0001601	1	3%
Cases with laryngomalacia requiring surgery		*1*	-
Dysphagia	HP:0002015	2	6%
Teeth (any)		**13**	**41%**
Malocclusion	HP:0000689	1	3%
Dental Crowding	HP:0000678	2	6%
Cases with dental crowding requiring surgery		*1*	-
Abnormal Number or Size of Teeth (any)	-	**5**	**16%**
Microdontia	HP:0000691	2	6%
Hyperdontia	HP:0011069	2	6%
Hypodontia	HP:0009804	1	3%
Abnormal Dentition (any)	-	**9**	**28%**
General (e.g., cavities, alignment)	HP:0000164	6	19%
Dental caries	HP:0000670	3	9%
Dental enamel	HP:0000682	1	3%
Cardiovascular		**16**	**50%**
Rhythm		**3**	**9%**

SVT at Delivery	HP:0004755	1	3%
Syncope	HP:0001279	2	6%
Cases with syncope diagnosed with POTS		*1*	*3%*
Structural		**15**	**47%**
Murmur	HP:0030148	7	22%
Complex congenital cardiovascular disease (any)	-	**8**	**25%**
Cases requiring surgery		*4*	-
Abnormal vascular (AVM)	HP:0002597	1	3%
Hypoplastic right heart	HP:0010954	1	3%
Patent ductus arteriosus (PDA)	HP:0011648	2	6%
Pulmonary artery stenosis	HP:0004415	1	3%
Pulmonary atresia artery	HP:0004935	1	3%
Pulmonary atresia valve	HP:0010882	1	3%
Pulmonary valve stenosis	HP:0001642	1	3%
Tricuspid stenosis	HP:0010446	1	3%
Ventral septal defect	HP:0001629	2	6%
Respiratory		**8**	**25%**
Frequent Infection	HP:0002205	1	3%
Recurrent Bronchitis	HP:0002837	1	3%
Asthma	HP:0002099	6	19%
Sleep		**10**	**31%**
Sleep Apnea	HP:0010535	1	3%
Sleep Disturbance (any)	HP:0002360	**10**	**31%**
Difficulty initiating sleep	-	5	16%
Difficulty maintaining sleep	-	6	19%
Sleep walking	HP:0025236	1	3%
Gastrointestinal		**26**	**81%**
Nausea	HP:0002018	1	3%
Redundant Colon	-	1	3%
Abdominal Pain (any)	-	**2**	**6%**
General abdominal pain	HP:0002027	1	3%
Episodic abdominal pain	HP:0002574	1	3%
Failure to Thrive in Infancy	HP:0001531	3	9%
Feeding Problems Beyond Infancy	HP:0011968	5	16%
Cases requiring feeding tube placement		*3*	-
Constipation	HP:0002019	13	41%
Failure to Thrive Beyond Infancy	HP:0001508	13	41%
Reflux	HP:0002020	16	50%
Feeding Problems in Infancy (e.g., poor latch, restrictive food preferences)	HP:0008872	19	59%
Renal/Genitourinary		**9**	**28%**
Renal Tubular Acidosis (RTA)	HP:0000787	1	3%
Vesicoureteral Reflux (VUR) with recurrent urinary tract infections (UTI)	HP:0000076	1	3%
Abnormal Penis (males only, any)	-	**2**	**10%**
		2	-

Cases with abnormal penis requiring surgery			
Cryptorchidism	HP:0000028	1	5%
Hooded prepuce	HP:0000036	1	5%
Hypospadias	HP:0003244	2	10%
Enuresis (any)	-	**7**	**22%**
General	HP:0000805	8	25%
Nocturnal	HP:0010677	2	6%
Endocrine		**9**	**28%**
History of High Prolactin	-	1	3%
Obesity secondary to medication	-	1	3%
Polydipsia	HP:0001959	1	3%
Polyphagia or Hyperphagia	HP:0002591	1	3%
Weight Gain	HP:0004324	1	3%
Hypothyroidism	HP:0000821	3	9%
Short Stature	HP:0004322	3	9%
Hematologic		**4**	**13%**
Bruising	HP:0000978	2	6%
Anemia (e.g., megaloblastic, secondary to epistaxis)	HP:0001903	2	6%
Musculoskeletal (any)		**8**	**25%**
Abnormal Thumb Phalanx	HP:0009602	1	3%
Limb Pain	HP:0009763	1	3%
Vertebral Fusion	HP:0002948	1	3%
Joint Laxity	HP:0001388	2	6%
Joint Pain	HP:0002829	2	6%
Joint Stiffness	HP:0001376	3	9%
Dermatologic		**11**	**34%**
Accessory Nipple	HP:0002558	1	3%
Livedo Reticularis	HP:0000965	1	3%
Striae Distensae	HP:0001065	1	3%
Vitiligo	HP:0001045	1	3%
Eczema	HP:0000964	4	13%
Keratosis Pilaris	HP:0032152	4	13%
Allergy/immunology		**9**	**28%**
Allergies			
Drug Allergy	HP:0410323	1	3%
Food Allergy	HP:0500093	4	13%
Seasonal Allergies	HP:0012395	5	16%
Deficiencies			
IGA Deficiency	HP:0002720	1	3%
IGG Deficiency	HP:0004315	1	3%
Neurological (any)		**18**	**56%**
Abnormal Pain Sensation (e.g., does not feel pain)	HP:0010832	1	3%
Abnormal Muscle Bulk	HP:0030236	1	3%
Tremors	HP:0001337	1	3%
Generalized Hypotonia	HP:0001290	2	6%
Tics or Movement Disorder	HP:0100033	2	6%

Seizures (any)	-	**4**	**13%**
Atonic	HP:0010819	1	3%
Febrile	HP:0002373	2	6%
Nocturnal	HP:0031951	1	3%
Unspecified	-	1	3%
Headache	HP:0002315	5	16%

Supplemental Table S3. Frequency of Physical Findings

Finding	Associated HPO Codes	n	%
Cardiovascular (any)		**2**	**6%**
Murmur (any)	-	2	6%
General	HP:0030148	1	3%
Systolic	HP:0031664	1	3%
Musculoskeletal (any)		**27**	**84%**
Axial (any)		**15**	**47%**
Long Neck	HP:0000472	2	6%
Scoliosis	HP:0002650	2	6%
Chest Deformities (any)	**-**	**13**	**41%**
Asymmetric Chest	HP:0001555	2	6%
Pectus Carinatum	HP:0000768	3	9%
Pectus Excavatum	HP:0000767	8	25%
Extremity – Upper (any)		**15**	**47%**
Fetal Fingertip Pads	HP:0001212	1	3%
Prominent Interphalangeal Epiphyses	HP:0010231	1	3%
Ulnar Deviation	HP:0009465	1	3%
Short Finger	HP:0009237	2	6%
Abnormal Palmar Crease (any)	-	3	9%
General	HP:0010490	2	6%
Single transverse	HP:0000954	1	3%
Tapered Finger	HP:0001182	4	13%
Long, Thin Finger (any)	-	**8**	**25%**
Long fingers	HP:0100807	4	13%
Thin fingers	HP:0001238	3	9%
Extremity – Lower (any)		**23**	**72%**
Genu Recurvatum	HP:0002816	1	3%
Small Feet	HP:0001773	1	3%
Leg Asymmetry	HP:0100559	2	6%
Long Toe	HP:0010511	2	6%
Abnormal Toes (any)	-	**9**	**28%**
Big hallux	HP:0001844	1	3%
Broad hallux	HP:0010055	1	3%
Broad hallux phalanx	HP:0010059	1	3%
Hallux valgus	HP:0001822	1	3%
Overlapping toes	HP:0001845	2	6%
Short phalanx of 2nd toe	HP:0010431	1	3%
Laterally curved 2nd toe	HP:0010319	2	6%
Medially curved 3rd toe	HP:0010320	2	6%
Curved 4th toe	HP:0010321	1	3%
Medial Rotation Medial Malleolus	HP:0008132	10	31%
Pes Planus	HP:0001763	10	31%
Dermatologic (any)		**11**	**34%**
Abnormal Fingernail	HP:0001597	1	3%
Axillary Freckling	HP:0000997	1	3%

Epidermal Nevus	HP:0010816	1	3%
Hyperpigmented Papule	HP:0025473	1	3%
Inguinal Freckling	HP:0030052	1	3%
Livedo Reticularis	HP:0000965	1	3%
Vitiligo	HP:0001045	1	3%
Abnormal Toenail (any)	-	**2**	**6%**
Hypoplastic	HP:0001800	1	3%
Dystrophic	HP:0001810	1	3%
Café-au-lait Macules	HP:0000957	2	6%
Dermal Translucency	HP:0010648	2	6%
Nevi or Macule (any)	-	**3**	**9%**
Hyper melanotic macule	HP:0001034	1	3%
Macule	HP:0012733	1	3%
Nevi	HP:0001054	1	3%
Neuro (any)		**9**	**28%**
General (any)		**3**	**9%**
Speech articulation difficulty	HP:0009088	1	3%
Hyperreflexia	HP:0001347	2	6%
Muscle (any)		**3**	**9%**
Abnormality of muscle size (decreased muscle bulk)	HP:0030236	1	3%
Hypotonia	HP:0001276	1	3%
Hypertonicity	HP:0001290	1	3%

Note. Numbers in bold face type represent a count of *any* instance in the category for each case (total possible = 32).

Supplemental Table S4: Findings from T1- and T2-weighted MRI scans in N = 24 patients with 3q29-deletion syndrome.

Participant	Demographic information	Posterior fossa structure	Abnormality
1	4 y/o, white (non-Hispanic) male	Normal	
2	6 y/o, white (non-Hispanic) male	Abnormal	Global cerebellar hypoplasia
3	6 y/o, white (non-Hispanic) male	Abnormal	Subtle cerebellar hemispheric hypoplasia
4	6 y/o, white (non-Hispanic) female	Abnormal	Retrocerebellar arachnoid cyst
5	6 y/o, white (non-Hispanic) female	Abnormal	Retrocerebellar arachnoid cyst
6	7 y/o, white (non-Hispanic) male	Abnormal	Cerebellar vermis hypoplasia
7	8 y/o, white (non-Hispanic) male	Normal	
8	8 y/o, white (non-Hispanic) male	Normal	
9	9 y/o, white (non-Hispanic) female	Normal	
10	10 y/o, white (non-Hispanic) male	Abnormal	Cerebellar vermis hypoplasia
11	10 y/o, more than one race (non-Hispanic) female	Abnormal	Retrocerebellar arachnoid cyst

12	12 y/o, more than one race (non-Hispanic) male	Abnormal	Cerebellar hemispheric hypoplasia
13	14 y/o, white (non-Hispanic) male	Abnormal	Cerebellar hemispheric hypoplasia
14	15 y/o, white (non-Hispanic) female	Normal	
15	15 y/o, white (non-Hispanic) male	Abnormal	Retrocerebellar arachnoid cyst, Cerebellar hemispheric hypoplasia
16	16 y/o, white (non-Hispanic) male	Normal	
17	17 y/o, white (non-Hispanic) male	Abnormal	Cerebellar vermis hypoplasia
18	18 y/o, white (Hispanic) male	Abnormal	Retrocerebellar arachnoid cyst, Cerebellar vermis hypoplasia
19	21 y/o, white (non-Hispanic) female	Abnormal	Retrocerebellar arachnoid cyst, Cerebellar hemispheric hypoplasia
20	21 y/o, white (non-Hispanic) female	Abnormal	Retrocerebellar arachnoid cyst, Subtle cerebellar vermis hypoplasia
21	24 y/o, white (non-Hispanic) male	Abnormal	Cerebellar vermis hypoplasia
22	27 y/o, white (non-Hispanic) female	Abnormal	Subtle cerebellar vermis hypoplasia
23	34 y/o, white (non-Hispanic) female	Abnormal	Cerebellar vermis hypoplasia, Cerebellar hemispheric hypoplasia

24	39 y/o, white (non-Hispanic) male	Normal

Supplemental Table S5: Demographic characteristics of patients with abnormal (left) and normal (right) posterior fossa structure.

Participants with an abnormal posterior fossa finding (*N* = 17; 70.8%)		**Participants with a normal posterior fossa finding** (*N* = 7; 29.2%)	
Sex		**Sex**	
Male	10 (41.7%)	Male	5 (20.8%)
Female	7 (29.2%)	Female	2 (8.3%)
Age		**Age**	
Mean (SD)	14.9 (8.4) years	Mean (SD)	14.1 (11.7) years
Median	14 years	Median	9 years
Range	6 – 34 years	Range	4 – 39 years
Ethnicity		**Ethnicity**	
Non-Hispanic	16 (66.7%)	Non-Hispanic	7 (29.2%)
Hispanic	1 (4.2%)		
Race		**Race**	
White	15 (62.5%)	White	7 (29.2%)
More than one race	2 (8.3%)		

Table S6: Results of Neurological Exam for 23 individuals with 3q29 deletion syndrome

	Fine Finger Movement	Rapid Alternating Movement	Heel to Shin Task	Finger to Nose Task	Tandem Walk
normal/score 0 (%)	6 (26%)	4 (17%)	6 (32%)	14 (61%)	11 (55%)
Mild/score 1 (%)	9 (39%)	7 (30%)	9 (47%)	8 (35%)	6 (30%)
moderate/score 2 (%)	8 (35%)	11 (48%)	4 (21%)	1 (4%)	3 (15%)
severe/score 3 (%)	0 (0%)	1 (4%)	0 (0%)	0 (0%)	0 (0%)
not normal (%)	17 (74%)	19 (83%)	13 (68%)	9 (39%)	9 (45%)

Medical History (~25 minutes)

These questions ask about your child's medical history. Please do your best to answer all applicable questions. Overall, these questions will take about 20-30 minutes to complete.

You can save and return to these questions at any time by clicking the Save and Return button at the bottom of the survey. Unsaved responses will have to be re-entered.

For questions, please email melissa.murphy@emory.edu or call (404) 727-3446.

Thank you!

Name of person completing this survey: _____

Relation to person with 3q29
- ◯ biological parent
- ◯ step-parent
- ◯ grandparent
- ◯ self
- ◯ other

Please describe your relation to the person with 3q29 _____

Please answer the following questions in reference to the person with 3q29.

When did you or your primary care provider first suspect a problem? _____

What current questions or concerns do you have about your child? _____

Is your child adopted?
- ◯ Yes
- ◯ No

Pregnancy History (for the pregnancy of the person with 3q29)

Mother's age at delivery? _____

Father's age at delivery? _____

The pregnancy was confirmed by:
- ◯ blood test
- ◯ urine test

At how many weeks was the pregnancy confirmed? _____

What number pregnancy was this for the mother?
- ◯ 1st
- ◯ 2nd
- ◯ 3rd
- ◯ 4th
- ◯ 5th
- ◯ Other

When did the mother begin prenatal care?

- ◯ 1st trimester
- ◯ 2nd trimester
- ◯ 3rd trimester
- ◯ No prenatal care

Please answer the following Yes/No questions about the pregnancy. If yes, please provide detail below.

	Yes	No	Don't Know
1. Prenatal vitamins?	◯	◯	◯
2. Medications (prescription)?	◯	◯	◯
3. Medications (over-the-counter)?	◯	◯	◯
4. Smoking?	◯	◯	◯
5. Alcohol (beer, liquor, wine)?	◯	◯	◯
6. Street drugs?	◯	◯	◯
7. Illness/infection?	◯	◯	◯
8. Bleeding?	◯	◯	◯
9. Rash?	◯	◯	◯
10. Fever?	◯	◯	◯
11. Diabetes?	◯	◯	◯
12. High blood pressure?	◯	◯	◯
13. Thyroid problems?	◯	◯	◯
14. X-rays/radiation?	◯	◯	◯
15. Premature labor?	◯	◯	◯
16. Hospitalization (do not count the delivery/birth)?	◯	◯	◯
17. Abnormal growth of baby?	◯	◯	◯
18. Other concerns?	◯	◯	◯

Please provide details regarding prenatal vitamins:

_____

Please provide details regarding presciption medications during pregnancy:

_____

Please provide details regarding over-the-counter medications during pregnancy:

_____

Please provide details regarding smoking during pregnancy:

_____

Please provide details regarding alcohol (beer, liquor, wine) consumption during pregnancy:

_____

Please provide details regarding street drug usage during pregnancy:

Please provide details regarding illness/infection during pregnancy:

Please provide details regarding bleeding during pregnancy:

Please provide details regarding rash during pregnancy:

Please provide details regarding fever during pregnancy:

Please provide details regarding diabetes during pregnancy:

Please provide details regarding high blood pressure during pregnancy:

Please provide details regarding thyroid problems during pregnancy:

Please provide details regarding X-rays/radiation during pregnancy:

Please provide details regarding premature labor:

Please provide details regarding hospitalizations during pregnancy (do not count the delivery/birth):

Please provide details regarding abnormal growth of baby:

Please provide details regarding other concerns:

Please answer the following Yes/No questions regarding testing that may have been done during the pregnancy.

Screening

	Yes	No	Don't Know
First Trimester Screen (ultrasound of baby's neck/Nucal Translucency/NT measurement plus blood work)	◯	◯	◯
Second Trimester Screen (Triple Screen, Quad Screen, AFP Test)	◯	◯	◯

Diagnostic Testing

	Yes	No	Don't Know
Chronic Villus Sampling (CVS)	◯	◯	◯
Amniocentesis	◯	◯	◯

Other

	Yes	No	Don't know
Glucose Tolerance Test	◯	◯	◯
Routine Ultrasound	◯	◯	◯
Specialized Ultrasound	◯	◯	◯
Other testing	◯	◯	◯

Please explain other testing that may have been done during the pregnancy: _____

Were any of the screening, diagnostic, or other tests ABNORMAL? If YES, please explain: _____

At how many weeks were the baby's first movements felt? _____

Were the baby's movements normal during the pregnancy?
◯ Yes
◯ No

Mother's total weight gain during pregnancy (in pounds): _____

Birth History (for the person with 3q29)

Due date:	_____
Date delivered:	_____
The child was born:	◯ Early ◯ On time ◯ Late
By how many weeks?	_____
Birth hospital (if not in GA, please include the state):	_____
Was the labor:	◯ Spontaneous (happened on its own) ◯ Induced
Please explain the reason for the induced birth and the method used (i.e. doctor broke your water, pitocin, etc.) if known:	_____
How was the child delivered?	◯ Vaginal ◯ C-section
Please explain the reason why a C-section was performed (i.e. previous child born that way, failure to progress, etc.):	_____
Was the baby born head first?	◯ Yes ◯ No ◯ I don't know
Baby's weight (in pounds):	_____
Baby's length (inches):	_____
Baby's head size (inches):	_____
Were there complications with the delivery?	◯ Yes ◯ No
Please list complications with the delivery:	_____
Were there any problems right after birth (i.e. need to go to the NICU, breathing problems, jaundice, etc.)?	◯ Yes ◯ No

Please describe problems right after birth:

_____

Did the baby have any feeding difficulties?

◯ Yes
◯ No

Please describe any feeding difficulties:

_____

Was your child born with any birth defects? (i.e. club foot, cleft lip and/or cleft palate, heart defects, extra fingers, etc.)?

◯ Yes
◯ No

Please describe birth defects:

_____

After the baby was born, how did he/she feed?

◯ Breast
◯ Bottle
◯ Other

Please describe how he/she fed:

_____

At how many days was your baby discharged home?

_____

Past Medical History (for the person with 3q29)

Please answer the following Yes/No questions about possible tests/procedures/etc. that your child may have had. If your child has had one of these things, please provide more detail in the box below (When? Why? Where? Results?).

General

	Yes	No
Had a formal eye examination with ophthalmology?	◯	◯
Had a formal hearing examination?	◯	◯
Been hospitalized overnight?	◯	◯
Had surgery?	◯	◯
Currently taking any medications?	◯	◯
Been tested for allergies?	◯	◯
Immunizations up to date?	◯	◯

Please comment on the formal eye examination your child had:

_____

Please comment on the formal hearing examination your child had:

_____

Please comment on the overnight hospital stay your child had:

_____

Please comment on the surgery your child had:

_____

Please comment on the medications your child is currently taking:

_____

Please comment on the allergy test(s) your child had:

_____

Please comment on your child's immunization history:

_____

Genetic

Ever had genetic testing?

◯ Yes
◯ No

Please comment on the genetic testing your child had:

_____

Imaging

	Yes	No
Had an MRI of the brain?	◯	◯
Had an MRI of the kidney?	◯	◯
Had an MRI of the heart?	◯	◯
Had a CT scan of the brain?	◯	◯
Had a CT of the kidney?	◯	◯
Had a CT of the heart?	◯	◯
Had an ultrasound of the brain?	◯	◯
Had an ultrasound of the kidney?	◯	◯
Had an ultrasound of the heart (echocardiogram)?	◯	◯
Had an X-ray of the brain?	◯	◯
Had an X-ray of the kidney?	◯	◯

Had an X-ray of the heart?	○	○
Had any other special procedures (i.e. EEG, swallow study, etc.)?	○	○

Please comment on the MRI of the brain your child had:

_____

Please comment on the MRI of the kidney your child had:

_____

Please comment on the MRI of the heart your child had:

_____

Please comment on the CT scan of the brain your child had:

_____

Please comment on the CT scan of the kidney your child had:

_____

Please comment on the CT scan of the heart your child had:

_____

Please comment on the ultrasound of the brain your child had:

_____

Please comment on the ultrasound of the kidney your child had:

_____

Please comment on the ultrasound of the heart (echocardiogram) your child had:

_____

Please comment on the X-ray of the brain your child had:

_____

Please comment on the X-ray of the kidney your child had:

_____

Please comment on the X-ray of the heart your child had:

_____

Please comment on any other special procedures (i.e. EEG, swallow study, etc.) that your child has had:

_____

Does your child have any significant problems with any of the following?

	Yes	No
Unusual weight gain or loss	◯	◯
Eye/vision	◯	◯
Hearing	◯	◯
Ears/Nose/Mouth/Throat	◯	◯
Teeth	◯	◯
Lungs/Breathing	◯	◯
Heart/Veins/Arteries/Circulation	◯	◯
Stomach/Intestines/Bowels	◯	◯
Kidney/Bladder/Genitals	◯	◯
Bones/Muscles (pain, weakness, abnormalities, etc.)	◯	◯
Joint pains/Swelling/Stiffness	◯	◯
Skin/Hair/Nails	◯	◯
Easy bruising/Bleeding or poor wound healing	◯	◯
Headaches/Seizures	◯	◯
Loss of balance or coordination	◯	◯
Loss of developmental skills	◯	◯
Sleep disturbances/Problems	◯	◯
Behavior/Psychological Problems	◯	◯
Growth	◯	◯
Heat or cold intolerance	◯	◯
Delays or problems with puberty	◯	◯
Hormones	◯	◯
Other	◯	◯

Please describe any significant problems with your child's unusual weight gain or loss:

_____

Please describe any significant problems with your child's eyes/vision:

_____

Please describe any significant problems with your child's hearing:

_____

Please describe any significant problems with your child's ears/nose/mouth/throat:

_____

Please describe any significant problems with your child's teeth:

_____

Please describe any significant problems with your child's lungs/breathing:

Please describe any significant problems with your child's heart/veins/arteries/circulation:

Please describe any significant problems with your child's stomach/intestines/bowels:

Please describe any significant problems with your child's kidney/bladder/genitals:

Please describe any significant problems with your child's bones/muscles (pain, weakness, abnormalities, etc.):

Please describe any significant problems with your child's joint pain/swelling/stiffness:

Please describe any significant problems with your child's skin/hair/nails:

Please describe any significant problems with your child's easy bruising/bleeding or poor wound healing:

Please describe any significant problems with your child's headaches/seizures:

Please describe any significant problems with your child's loss of balance or coordination:

Please describe any significant problems with your child's loss of developmental skills:

Please describe any significant problems with your child's behavioral/psychological problems:

Please describe any significant problems with your child's sleep disturbances/problems:

Please describe any significant problems with your child's growth:

Please describe any significant problems with your child's heat or cold intolerance:

_____

Please describe any significant problems with your child's delays or problems with puberty:

_____

Please describe any significant problems with your child's hormones:

_____

Diet/Feeding History

Please describe any other significant problems:

_____

Please describe any past problems with your child's diet or feeding:

_____

Please describe any current problems with your child's diet or feeding:

_____

Parental Height

Biological father's height:

_____

Biological mother's height:

_____

Handedness

For each immediate biological family member of the person with 3q29, please indicate hand preference:

	right hand dominant	left hand dominant	uses both hands equally (ambidextrous)	unknown
Person with 3q29	◯	◯	◯	◯
Biological Mother	◯	◯	◯	◯
Biological Father	◯	◯	◯	◯
Sibling 1	◯	◯	◯	◯
Sibling 2	◯	◯	◯	◯

Does the child have more than 2 siblings?

◯ Yes
◯ No

Please list each additional sibling and his/her handedness.

_____

Early Development

WHEN did you or your doctor first become concerned about your child's development?

_____

If there were any concerns about your child's development, HOW were these concerns noticed?

_____

Pediatrician/Primary Care Physician

***If your child has changed pediatrician/primary care provider, please list the most recent provider.**

What is the name of your pediatrician/primary care physician?

_____

If you haven't already, are you willing to sign a release to allow us to contact this provider to request medical records?

◯ Yes
◯ No

Practice name:

_____

Practice address:

_____

Practice phone number:

_____

Practice fax:

_____

Other than a pediatrician/primary care physician, what doctors is your child ACTIVELY seeing?

Name of Doctor 1

_____

Specialty (i.e. neurology, cardiology, GI, etc.)

_____

Reason your child is seen

_____

How often child sees this doctor (i.e. once a year, every 3 months, etc.)

_____

Practice name:

_____

Practice address:

_____

Practice phone number:	_____
Enter other doctor	◯ Yes ◯ No
Name of Doctor 2	_____
Specialty (i.e. neurology, cardiology, GI, etc.)	_____
Reason your child is seen	_____
How often child sees this doctor (i.e. once a year, every 3 months, etc.)	_____
Practice name:	_____
Practice address:	_____
Practice phone number:	_____
Enter other doctor	◯ Yes ◯ No
Name of Doctor 3	_____
Specialty (i.e. neurology, cardiology, GI, etc.)	_____
Reason your child is seen	_____
How often your child sees this doctor (i.e. once a year, every 3 months, etc.)	_____
Practice name:	_____
Practice address:	_____
Practice phone number:	_____
Enter other doctor	◯ Yes ◯ No

Name of Doctor 4

_____

Specialty (i.e. neurology, cardiology, GI, etc.)

_____

Reason your child was seen

_____

How often child sees this doctor (i.e. once a year, every 3 months, etc.)

_____

Practice name:

_____

Practice address:

_____

Practice phone number:

_____

Enter other doctor

◯ Yes
◯ No

Please list other doctors your child is ACTIVELY seeing (include specialty, frequency, and contact information):

_____

Other than a pediatrician/primary care physician, what doctors has your child seen IN THE PAST?

Name of Doctor 1

_____

Specialty (i.e. neurology, cardiology, GI, etc.)

_____

Reason your child was seen

_____

Date of last visit with this specialist

_____

Enter other doctor

◯ Yes
◯ No

Name of Doctor 2

_____

Specialty (i.e. neurology, cardiology, GI, etc.)

_____

Reason your child was seen

_____

Date of last visit with this specialist

_____

Enter other doctor

◯ Yes
◯ No

Name of Doctor 3

_____

Specialty (i.e. neurology, cardiology, GI, etc.)

_____

Reason your child was seen

_____

Date of last visit with this specialist

_____

Enter other doctor

◯ Yes
◯ No

Name of Doctor 4

_____

Speciality (i.e. neurology, cardiology, GI, etc.)

_____

Reason your child was seen

_____

Date of last visit with this specialist

_____

Enter other doctor

◯ Yes
◯ No

Please describe other doctors your child has seen in the PAST (include specialty and frequency):

_____

How old was your child when he/she began:

Rolling over?

_____

Sitting alone?

_____

Pulling to stand?

_____

Crawling?

_____

Cruising?

_____

Walking alone?

_____

First word?

_____

First sentences?

_____

Toilet trained?

_____

Has your child lost any skills that he/she previously mastered (regression)?

◯ Yes
◯ No

Please describe any regression:

_____

School Information

Does your child currently attend school or day care?

◯ Yes
◯ No

What is the name of the school/daycare?

_____

Grade (if applicable)?

_____

Does your child attend special classes or need special help?

◯ Yes
◯ No

Please describe special classes your child attends or special help he/she needs (For example, what subjects does he/she need help in? Is he/she in an inclusion class or self-contained class?):

_____

If not already provided, please upload a copy of your child's most recent IEP (if possible):

Does your child receive any of the following?

	Yes	No
Physical therapy	◯	◯
Occupational therapy	◯	◯
Speech therapy	◯	◯

Other therapy	◯	◯

How often does your child receive physical therapy?

_____

How often does your child receive occupational therapy?

_____

How often does your child receive speech therapy?

_____

Please list other therapy your child receives and how often:

_____

Does your child have any behavioral problems?

◯ Yes
◯ No

Please describe any behavioral problems:

_____

Do you feel that your child's language skills are where they should be for your child's age?

◯ Yes
◯ No

Please describe your child's language skills for his/her age:

_____

Has your child ever had IQ testing or a formal developmental assessment?

◯ Yes
◯ No

When did your child receive IQ testing or a formal development assessment? And what were the results?

_____

If not already provided, please upload a copy of the results from the most recent evaluation (if possible):